that needs further study. The potential for posterior dislocation of the donor corneal disc in aphakic eyes must be taken into consideration. Longer-lasting, higher-buoyancy gases (such as SF₆) could be used as alternatives to air, but the possible toxic effect to endothelial cells should be elucidated.

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Cherry Red Spot in Sialidosis (Mucolipidosis Type I)

The differential diagnosis of a cherry red spot in the macula includes central retinal artery occlusion and metabolic storage diseases such as Tay-Sachs disease, Sandhoff disease, Niemann-Pick disease, Fabry disease, Gaucher disease, and sialidosis. We report a case of an adolescent who, at a routine ophthalmic examination, was found to have a cherry red spot in the maculae of both eyes. Laboratory investigation results showed that the patient had mucolipidosis type I, which is a rare lysosomal storage disease with clinical and histologic findings similar to the mucopolysaccharidoses and the sphingolipidoses.

Report of a Case. A 14-year-old white boy complained of difficulty seeing the blackboard at school. A screening eye examination found decreased distance vision in both eyes. He was of normal intelligence and his medical history was significant only for scoliosis and seasonal allergies. Visual acuity corrected to 20/20 OU. Results of a dilated ophthalmoscopic examination revealed a cherry red spot in both maculae (Figure). Fluorescein angiography demonstrated hypofluorescence around the foveal area in the midvenous phase (eFigure 1, available online at http://www.archophthalmol.com). There was relative hyperfluorescence in the foveal area, but this was believed to represent a normal fluorescein pattern, rather than pigment epithelial disease. Optical coherence tomography showed increased reflectivity of the inner retinal layers, corresponding to the hypofluorescence on the fluorescein angiogram (eFigure 2). Because
of the patient’s normal visual acuity, symmetric changes, and lack of symptoms, we suspected a metabolic storage disorder. Laboratory test results showed normal levels of β-galactosidase, arylsulfatase A, hexosaminidase A and B, and β-galactocerebroside. However, sialic acid levels were elevated in the urine. Examination results of a skin biopsy specimen revealed a fibroblast α-neuraminidase level of 0.5 nmoles/h per milligram of protein (reference range, 15.0-30.0 nmoles/h per milligram of protein). Based on these findings, a diagnosis of sialidosis (mucolipidosis type I) was made.

Comment. Sialidosis (mucolipidosis type I) is a rare inherited lysosomal storage disease characterized by deficiency of α-N-acetyleneuraminidase (sialidase) in leukocytes and cultured fibroblasts. This results in intracellular storage of excess sialyloligosaccharides and is histologically observed as abnormal vacuolization of various cell types. Two major phenotypes of mucolipidosis exist: type I or the cherry red spot myoclonus syndrome, and a more severe infantile form, type II. Sialidosis has an autosomal recessive pattern of inheritance, and the gene has been localized on chromosome 6p21.2 It occurs in 1 of every 2 200 000 live births. Patients with type II disease have somatic changes, characterized by coarse facies, hepatolomegaly, bony changes of dysostosis multiplex, and developmental delay. Patients with type II disease usually die within the first 2 years (congenital form) or by the second decade (infantile form) of life.3 By contrast, patients with type I disease are less severely affected and typically develop symptoms of myoclonic epilepsy, visual problems, and ataxia in the second or the third decade of life.4 Macular cherry red spots are always present and, therefore, sialidosis should be included in the differential diagnosis of a cherry red macula in this clinical setting. Diagnostic evaluation for a patient with a cherry red spot in the macula not due to arterial occlusion should include a genetic history and an appropriate laboratory workup to confirm the underlying cause.

Conclusions. We herein present a rare case of a 14-year-old boy with mucolipidosis type I who had cherry red spots in the macula of both eyes. The patient had minimal symptoms and visual acuity correctable to 20/20 OU. Of interest in this case are the fluorescein angiogram and optical coherence tomogram findings. The fluorescein angiogram shows blocked fluorescence surrounding the fovea throughout the angiogram (eFigure 1). The optical coherence tomogram shows increased reflectivity in the ganglion cell layer corresponding to the blocked fluorescence (eFigure 2). Although pathological confirmation is lacking in this case, a previous autopsy report of a patient with mucolipidosis type I demonstrated diffuse intracytoplasmic accumulation of lipofuscinlike pigment in the cerebral neurons.5 This leads us to speculate that the ophthalmoscopic changes observed in our patient are due to an accumulation of sialyloligosaccharides in the inner layers of the retina.

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Macular Retinal Detachment in Hallermann-Streiff Syndrome

Hallermann-Streiff syndrome (also known as oculomandibulofacial syndrome) is a rare syndrome chiefly comprising facial and ocular abnor-